



# Long-Term Follow-up of Newborn Screening

## WHAT IS THE PUBLIC HEALTH ISSUE?

Phenylketonuria (PKU) is a genetic disorder that affects the body's ability to convert certain proteins found in foods. Newborn screening was implemented in the 1960s to provide early diagnosis of PKU. Identifying the disorder early allows dietary management to prevent the development of mental retardation in affected children. Since then, newborn screening programs have expanded to incorporate additional conditions such as hearing loss and disorders which affect a person's metabolism, blood, or hormones. Newborn screening programs have traditionally been limited to conditions that are serious, treatable, or controllable, influenced by age of diagnosis, and with a natural history that is understood.

In the 1990s, the technology of tandem mass spectrometry was introduced for population-based newborn screening. This technology allows for more accurate measurement of a broader range of conditions than was previously available. For some conditions, such as PKU, the benefits of newborn screening and early treatment are generally accepted. However, for other conditions there is little information on the long-term benefits from screening and intervention. Population-based tracking and follow-up studies of children identified through the latest technology are needed to assess the public health impact of newborn screening for many of the disorders identified with this technology. Ideally, this approach will include collection of information related to treatment options, treatment compliance, and long-term outcomes. The infrastructure developed for this assessment can also be used in the future for evaluation of new newborn screening programs.

## WHAT HAS CDC ACCOMPLISHED?

- Created hypothesis for assessing outcomes.
- Funded Iowa and a collaboration in Oregon and Idaho to develop a database system for long-term follow-up of newborn screening.
- Finalized research protocol for the collection and analysis of medical records data.
- Developed a database to assess the long-term effects of newborn screening.

## WHAT ARE THE NEXT STEPS?

- Adding component of standard developmental assessment for all children within the study group.
- Continuing medical record review and data collection.
- Expanding the protocol to additional sites and additional disorders.